Report To CLIAC on SACGT

Patricia Charache, M.D.

September, 12, 2002

SACGT Report
September 12, 2002

The Charter for SACGT expired in August.

- DHHS has decided not to renew the charter of SACGT

Format of This Presentation

- Summarize the premise and outputs of SACGT in areas in which its activities relate to issues that impact directly on quality of patient care laboratory practices.
- (The body of SACGT's activities involved areas not in CLIAC's purview.)

Background Premise

- Over 800 genetic tests now exist, (577 in CLIA approved labs, 368 in research labs). Most target rare genetic disorders; others are being developed.
- These tests have multiple uses, e.g. newborn screening, carrier screening, predictive testing, disease diagnosis or prognosis, pharmacogenetics.
- Some, especially predictive tests, raise sensitive medical, social, ethical, and legal issues.

SACGT Charter

- Advise the Secretary on all aspects of the development and use of genetic tests. Includes
  - safe and effective incorporation of genetic technologies into health care
  - assessing the effectiveness of existing and future measures for oversight of genetic tests, and
  - identifying research needs related to the Committee's purview.

Accomplishments

- **Recommendations By SACGT (7/00)**

  There is a need:

  - **To improve the oversight of genetic tests**
  - **For Federal legislation to prevent discrimination in insurance and employment**
  - **Study the effect of gene patents and licensure**
  - **Study further the issue of informed consent of third parties in human research subjects.**

Recommendations Pertaining to Adequacy of Oversight of Genetic Tests (Continued)

- The FDA should regulate laboratory developed genetic tests ("home brews"), using an innovative, flexible approach
- CLIA should be augmented to incorporate specific provisions for genetic testing laboratories
- Private-public collaborations are needed to ensure continued analysis of post market data

Definitions: (PC痴)

- Analytical Validity: Primarily concerned with ability to accurately measure a given analyte.
- Clinical validity: Ability to separate clinical disease from no disease or risk of disease through measuring that analyte.
- Clinical utility: Clinical validity plus full knowledge of test , including gene penetrance, etc.significance in populations to be tested.

Ongoing SACGT Considerations: Oversight

Who is responsible

Activity	IRB	CLIA	FDA
Research (development)	X		
Research, (validated analytically, clinically) limited patient reports	X	X	
Wide use patient reports, fully validated, +/-continued research	+/-	X	X

Ongoing SACGT Activities: Work Groups and Task Forces

- Pursued recommendation issues
- Established work groups for additional issues related to other aspects of testing
  - Education
  - IRB/Consent
  - Rare Diseases
  - Access
  - Data collection, clinical utility information

Education Work Group

- **Assess the adequacy of current efforts to advance genetics education of health professionals**
- **Year-long data gathering and fact finding; educational summit in Baltimore, May, 2002.**

<u>Issues:</u> For appropriate pre- and post-analytical aspects of testing, educated users are required. Laboratory Directors, IRB's, clinicians, others need knowledge base.

Consent/IRB Work Group

- **A brochure was developed to explain genetic testing and informed consent to the public**
- **White paper was under development on principles of informed consent, defining levels of consent, and consent recommendations for various types of genetic tests**

<u>Laboratory Issues:</u> Who decides level of consent What is the laboratory痴 role in assuring patient consent?

Rare Disease Testing Work Group

- **Definition of a rare genetic disease**
- **Developmental and practice incentives**
- **Special access issues**
- **Quality assurance and validation assistance for research laboratories testing for rare diseases.**

<u>Issues</u>: Limited test sites, mainly research labs , home brew tests if limited industrial interest, no proficiency tests, patent issues

Access Work Group Discussions

- Reimbursement for:
  - Test cost
  - Genetic education and counseling
  - Other professional services
  - Non-reimbursed laboratory costs
- Health care disparities
- Gene patents and licensing:
  - Value for industrial interest in development
  - Issue for access and quality assurance

Data Work Group

<u>Goal</u>: To improve knowledge of the disease and the clinical validity and utility of a test

- **Needs: Improved post market data collection, access to data, resources for data organization, and analysis. Both clinical and laboratory data are required**
- **Survey of HHS activities to advance knowledge of clinical validity and utility (translational research)**

<u>Lab Issues</u>: Who is to provide the data and how? Privacy? Cost? Definitions of a test, etc.

Additional Concerns Supportive of CLIAC's Reports

- Waived tests (of major concern as they apply to genetic testing because of pre- and post analytical considerations)
- CMS study of laboratories performing waived tests

Summary

SACGT recommendations and considerations:

- **Oversight functions, including FDA review of tests, template approach and enhanced CLIA**
- **Additional subject matter covered by work groups and task forces: Education, IRB/Consent, Rare Diseases, Access, Data Work Group**
- **Other issues: Patent issues; (Waived tests, CMS findings of Waived testing laboratories)**

Outstanding Issues

- Classification of laboratory oversight responsibilities, clarifying when CLIA applies to research facilities
- Provision of education/guidance documents for IRB's, and/or research laboratories interested in patient care
- Oversight of laboratory developed tests: CMS and deemed status organization feasible assessment instruments for analytical and clinical validation (not full clinical utility).

Outstanding Issues

- Informed consent issues, (check off box on lab requests?)
- Reimbursement for laboratory expenses associated with clinical user discussions.
- Education of Laboratory Directors and Technical Supervisors specific to genetic testing
- Consideration of result implications in test categorization decisions, e.g. waived vs. other .